Introduction

What is Newborn Screening?

Newborn Screening is a comprehensive public health program designed to prevent severe and potentially lethal outcomes from a variety of congenital disorders. The newborn screening system includes:

– Education to Hospitals, Providers and Families
– Screening of all newborn infants
– Follow-up of abnormal findings
– Evaluation, Diagnosis and Treatment
– Lifelong Management of identified disorders

All infants born in South Carolina are required by law to be screened in accordance with the regulation promulgated by the Board of the Department of Health and Environmental Control (DHEC). This regulation is further defined by Official Departmental Instructions that specify the roles and responsibilities of each entity involved in the newborn screening process.

A blood spot specimen is to be collected from each infant born in SC, preferably between 24 and 48 hours of age, and sent to the DHEC Public Health Laboratory (PHL) within 24 hours of collection. At present, infants are tested for select metabolic, immune, hormone/enzyme, and genetic disorders. The specific disorders on the test panel are included in this manual.

Purpose

The purpose of newborn screening is to identify infants at risk and in need of more definitive testing. **As with any laboratory screening test, both false positive and false negative results are possible. Initial screening test results are insufficient information to base definitive diagnosis or treatment.**

This manual uses terminology consistent with the American College of Medical Genetics (ACMG) report "*Newborn Screening: Towards a Uniform Screening Panel and System*," Genetic Med 2006; 8 (5) Supple: S12-S252. Tests for other disorders may be added in the future.

Select Disorders Table

The table below shows an estimate of the number of infants born with selected detectable newborn screening disorders in South Carolina:

Select Disorders	# Of Infants Diagnosed with a Newborn Screening Disorder in 2020
Hemoglobin Disorders	80
Hemoglobinopathy Traits & Carriers	2805
Congenital Hypothyroidism (CH)	19
Partial Biotinidase Deficiency	11
Cystic Fibrosis (CF)	10
MCAD	4
Congenital Adrenal Hyperplasia (CAH)	2
T-Cell Lymphopenia	3
CPT 1A	1
Classic Galactosemia	1
Metabolic Disorder Carriers	15

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- 2-methyl-3-OH-butyric Aciduria (2M3HBA)
- 3-hydroxy-3-methylglutaric aciduria (HMG)
- 3-methylglutaconic aciduria (3MGA)
- Multiple Carboxylase Deficiency (MCD)
- Glutaric Acidemia Type I (GA I)

- Fatty Acid Oxidation Disorders
  - Medium Chain Acyl CoA Dehydrogenase Deficiency (MCAD)
    - Medium chain ketoacyl CoA Thiolase deficiency (MCAT)
  - *Short Chain acyl CoA Dehydrogenase Deficiency* (*SCAD*)
  - Medium/Short Chain 3-OH acyl CoA Dehydrogenase Deficiency (M/SCHAD)
  - Dienoyl co-A Reductase Deficiency (DE RED)
  - Long Chain 3-OH acyl CoA Dehydrogenase Deficiency (LCHAD)
  - Trifunctional Protein Deficiency (TFP)
  - Very Long Chain acyl CoA Dehydrogenase Deficiency (VLCAD)
  - Glutaric acidemia type II (GA II)
  - Carnitine Palmitoyl transferase I Deficiency (CPT IA)
  - Carnitine Palmitoyl transferase II Deficiency (CPT II)
  - Carnitine/Acylcarnitine Translocase Deficiency (CACT)
  - Carnitine Uptake/Transport Defect (CUD)

- Endocrine and Enzyme Disorders
  - Congenital Hypothyroidism (CH)
  - Congenital Adrenal Hyperplasia (CAH)
  - Biotinidase Deficiency (BIOT)

- Hemoglobin Disorders
  - Sickle Cell Disease (Hgb SS)
  - Sickle C Disease (Hgb S/C)
  - Sickle ß Thalassemia (Hgb S/ß Th)
  - Various Hemoglobin Disorders and Traits
  - Sickle Cell Foundation Contacts in South Carolina

- Other Genetic Disorders
  - Cystic Fibrosis (CF)
  - Severe Combined Immunodeficiency (SCID)
    - T cell related immune disorders
  - Hearing Loss (HL)*
  - Critical Congenital Heart Defects (CCHD)*

- DHEC Newborn Screening Contact Information
- Newborn Screening Brochures and Forms
- Instructions for Completing NBS Collection Form - DHEC 1327
- Best Specimen Collection Timing by Disorder
- Criteria for Notification of Abnormal Results
  - Weekdays/ Monday holidays